FAM134B gene

family with sequence similarity 134 member B

Normal Function

The *FAM134B* gene provides instructions for making a protein important for the survival of sensory and autonomic nerve cells (neurons). Sensory neurons transmit pain, touch, and temperature sensations. Autonomic neurons help control involuntary functions of the body such as heart rate and blood pressure. Within neurons, the FAM134B protein is found in a structure called the Golgi apparatus, which is important for distribution of proteins to the cell. However, the function of the FAM134B protein is unknown. Studies have shown that neurons in which the FAM134B protein is absent die by a process of self-destruction called apoptosis.

Health Conditions Related to Genetic Changes

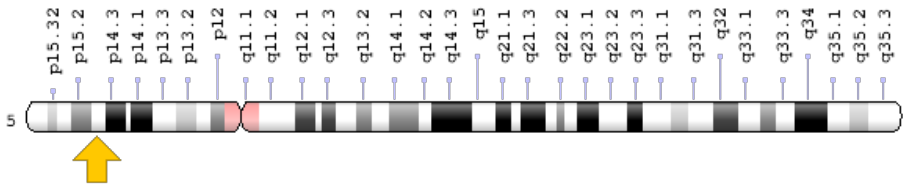
hereditary sensory and autonomic neuropathy type II

Mutations in the *FAM134B* gene are responsible for one type of hereditary sensory and autonomic neuropathy type II (HSAN2) called HSAN2B. People with HSAN2B lose the ability to feel pain or sense hot and cold. The *FAM134B* gene mutations may lead to an abnormally short and nonfunctional protein. The lack of FAM134B protein causes neurons to undergo apoptosis, which reduces the overall number of sensory and autonomic neurons. The loss of neurons results in the signs and symptoms of HSAN2B.

Chromosomal Location

Cytogenetic Location: 5p15.1, which is the short (p) arm of chromosome 5 at position 15.1

Molecular Location: base pairs 16,473,038 to 16,617,058 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- F134B_HUMAN
- FAM134B protein
- FAM134B protein isoform 1
- FAM134B protein isoform 2
- family with sequence similarity 134, member B
- FLJ20152
- FLJ22155
- FLJ22179
- JK1

Additional Information & Resources

GeneReviews

- Hereditary Sensory and Autonomic Neuropathy Type II
  https://www.ncbi.nlm.nih.gov/books/NBK49247

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28FAM134B%5BTIAB%5D%29+OR+%28JK1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- FAMILY WITH SEQUENCE SIMILARITY 134, MEMBER B
  http://omim.org/entry/613114

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FAM134B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RETREG1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25964
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/54463
- UniProt
  http://www.uniprot.org/uniprot/Q9H6L5

Sources for This Summary

- OMIM: FAMILY WITH SEQUENCE SIMILARITY 134, MEMBER B
  http://omim.org/entry/613114
- Kurth I, Pamminger T, Hennings JC, Soehendra D, Huebner AK, Rotthier A, Baets J, Senderek J, Topaloglu H, Farrell SA, Nürnberg G, Nürnberg P, De Jonghe P, Gal A, Kaether C, Timmerman V, Hübner CA. Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. Nat Genet. 2009 Nov;41(11):1179-81. doi: 10.1038/ng.464. Epub 2009 Oct 18.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19838196
- Murphy SM, Davidson GL, Brandner S, Houlden H, Reilly MM. Mutation in FAM134B causing severe hereditary sensory neuropathy. J Neurol Neurosurg Psychiatry. 2012 Jan;83(1):119-20. doi: 10.1136/jnnp.2010.228965. Epub 2010 Nov 28.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21115472
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3721196/
- Verpoorten N, De Jonghe P, Timmerman V. Disease mechanisms in hereditary sensory and autonomic neuropathies. Neurobiol Dis. 2006 Feb;21(2):247-55. Epub 2005 Sep 23. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16183296

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/FAM134B

Reviewed: May 2011
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services